



C2 gene

complement C2

Normal Function

The C2 gene provides instructions for making the complement component 2 protein. This protein helps regulate a part of the body's immune response known as the complement system. The complement system is a group of proteins that work together to destroy foreign invaders (such as bacteria and viruses), trigger inflammation, and remove debris from cells and tissues. When a foreign invader is detected, the complement pathway is turned on (activated) and the complement component 2 protein attaches (binds) to a similar protein called complement component 4. Together, these proteins form a complex called C3 convertase, which triggers further activation of the pathway, allowing the proteins of the complement system to participate in an immune response.

Health Conditions Related to Genetic Changes

age-related macular degeneration

complement component 2 deficiency

At least five mutations in the C2 gene have been found to cause complement component 2 deficiency. This disorder reduces the normal function of the immune system, resulting in an increased risk for infections and autoimmune disorders such as systemic lupus erythematosus (SLE). Autoimmune disorders occur when the immune system malfunctions and attacks the body's tissues and organs.

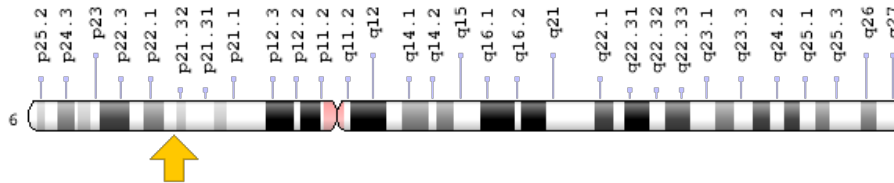
More than 90 percent of people with complement component 2 deficiency have a mutation that deletes 28 DNA building blocks (nucleotides) from the C2 gene. This mutation prevents the production of any complement component 2 protein. Without this protein to form C3 convertase, activation of the complement system is stalled. As a result, the complement system's ability to fight infections is diminished. It is unclear how complement component 2 deficiency leads to increased susceptibility to autoimmune disorders. Researchers speculate that the dysfunctional complement system is unable to distinguish what it should attack, and it sometimes attacks normal tissues, leading to autoimmunity. Alternatively, the dysfunctional complement system may perform partial attacks on invading molecules, which leaves behind foreign fragments that are difficult to distinguish from the body's tissues, so the complement system sometimes attacks the body's own cells. It is likely that other factors, both

genetic and environmental, play a role in the variability of the signs and symptoms of complement component 2 deficiency.

Chromosomal Location

Cytogenetic Location: 6p21.33, which is the short (p) arm of chromosome 6 at position 21.33

Molecular Location: base pairs 31,897,785 to 31,945,674 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARMD14
- C3/C5 convertase
- CO2
- complement component 2
- complement component C2

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): The Complement System and Innate Immunity
<https://www.ncbi.nlm.nih.gov/books/NBK27100/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28complement+C2%5BTIAB%5D%29+OR+%28complement+component+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- COMPLEMENT COMPONENT 2
<http://omim.org/entry/613927>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=C2%5Bgene%5D>
- HGNC Gene Family: Complement system
<http://www.genenames.org/cgi-bin/genefamilies/set/492>
- HGNC Gene Family: Sushi domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1179>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1248
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/717>
- UniProt
<http://www.uniprot.org/uniprot/P06681>

Sources for This Summary

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